

Public release date: 16-Jun-2007

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[European Society of Human Genetics](#)

Noninvasive screening in early pregnancy reduces Down's births by 50 percent

Nice, France: Non-invasive screening of pregnant women with ultrasound early in pregnancy, combined with maternal blood analysis, has reduced the number of children born in Denmark with Down Syndrome by 50%, a scientist will tell the annual conference of the European Society of Human Genetics today. Professor Karen Brødum-Nielsen, of the Kennedy Institute, Glostrup, Denmark, will say that another benefit of the introduction of this procedure in her country was a drop in the number of invasive pre-natal diagnostic procedures from 11% to approx. 6% of pregnancies.

In September 2004, Professor Brødum-Nielsen will tell the conference, the National Board of Health in Denmark recommended new guidelines for prenatal diagnosis. Previously this was restricted to pregnant women over 35 years of age, but since the implementation of the new guidelines it has been available to any woman who wants it.

The women were offered a measurement of nuchal translucency in the fetus by ultrasound. This test looks at thickness of the black space (fluid) in the neck area of the fetus. If there is more than the normal amount of fluid the risk of Down syndrome is increased. Likewise if there is a certain combination of serum markers in the maternal blood test, taken at the same time, there is the possibility of an increased risk of a chromosomal abnormality. The combined screening is carried out at 11 to 14 weeks of gestation.

Professor Brødum-Nielsen and her team looked at the effects of the new guidelines in 2004, 2005, and 2006, in 3 counties in Denmark with a total population of 1.1 million inhabitants, or about one-fifth of the population of the country. They compared these findings with national figures obtained from the Central Cytogenetic Registry, which confirmed the reduction in invasive procedures and the number of children born with Down syndrome at national level. When we looked further at the history of children born with Down Syndrome, we found that their mothers had declined the offer of screening, or had taken it up too late in pregnancy," she says. Another group had risk assessment that did not lead to invasive procedures

Women whose test results showed an elevated risk were offered an invasive procedure (chorionic villus sampling or amniocentesis) to definitely confirm or exclude the diagnosis of Down syndrome by chromosome analysis. We found that making non-invasive screening available to all pregnant women meant that the numbers of invasive procedures decreased by 40% between 2004 and 2006, says Professor Brødum-Nielsen. Although we have not yet studied the whole of the population, these numbers are significant enough to show that the new guidelines have been accepted by a great majority of Danish parents. However, there is a need for analysis of the psychosocial aspects, both as to the pre-test counselling and the women's attitudes, she says.

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